

ALBERTO LUIS ROSA

Investigador Principal CONICET

Director Laboratorio de Genética, Biología Celular y Molecular – IFEC – FCQ - UNC

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LINEAS DE INVESTIGACIÓN

Aspectos genéticos, bioquímicos, de biología celular y molecular de **levaduras**: microbiotas nativas de vides y mecanismos moleculares de resistencia a agentes tóxicos.

Bases moleculares y fisiopatogenia de **enfermedades neuromusculares hereditarias**: mecanismos genéticos de expresión del gen DUX4.

TITULOS DE GRADO

Licenciado en Química Biológica (curriculum de cinco años) (1983), Facultad de Ciencias Químicas, Universidad Nacional de Córdoba, Córdoba, Argentina

Magister en Biología Molecular y Microbiología (curriculum de dos años) (1985), INGEBI, SECYT, CONICET – UN La Plata, Buenos Aires, Argentina

Médico Cirujano (curriculum de seis años) (1995), Facultad de Ciencias Médicas, Universidad Nacional de Córdoba, Córdoba, Argentina

TITULOS DE POSGRADO

Doctor en Ciencias Químicas (1987), Facultad de Ciencias Químicas, Universidad Nacional de Córdoba. Tema: “Biología y Genética Molecular”. (área de investigación: genética, bioquímica, biología molecular y celular). Director: Dr. Hugo J.F. Maccioni.

Doctor en Medicina y Cirugía (2002), Facultad de Ciencias Médicas, Universidad Nacional de Córdoba. Tema: “Enfermedades hereditarias neurodegenerativas olivopontocerebelosas”. (área de investigación: genética humana, genómica, neuroquímica, biología molecular y celular). Director: Dr. Santiago Palacio.

TESINAS, TESIS Y POSTDOCRADOS DIRIGIDOS

TESINAS: Lic. R. Magni (1988), Biol. E. Temporini (1989), Biol. S. Haedo (1990), Biol. M. Alejandra Griffó (1991), Lic. G. Olalla (1996), Biol. I. Slavin (2000)

TESIS DE MAESTRIA: Bioq. A.R. Alfonso Pecchio (2002), Bioq. C. Castellán (2013)

TESIS DOCTORADO: Lic. M.R. Mautino (1995), Biol. E.D. Temporini (1995), Biol. S.D. Haedo (1995), Biol. G.A. Borioli (1998), Lic. Química J. Valdez (1999), Bioq. D. H. Folco (2003), Bioq. V. Kowaljow (2004), Bioq. E.D. Corona (2012), Lic. M.L. Raymond Eder (2021), Bioq. J. Quintero (2021), Bioq. S. Pagnoni (2021)

POST-DOCTORALES: Dra. M.E. Álvarez (1992-1994), Dr. J.L. Barra (1994-1995), Dra. C. Conde (1998-2001), Dra. D. Jacquelin (2010-2012), Dra. L. Gatica (2012-2014), Dra. M.L. Raymond Eder (2021-actual), Dra. S.M. Pagnoni (2021-actual)

SUBSIDIOS PARA INVESTIGACION CIENTIFICA

NACIONALES

Consejo Nacional de Investigaciones Científicas y Técnicas (CONICET)

Consejo de Investigaciones Científicas y Técnicas de Córdoba (CONICOR)

Fundación Antorchas

Secretaría de Ciencia y Técnica de Córdoba

Secretaría de Ciencia y Técnica de la Universidad Nacional de Córdoba

FONCYT de la Agencia Nacional de Promoción Científica y Técnica (ANPCYT)

Ciencia y Tecnología de la Universidad Católica de Córdoba

INTERNACIONALES

Third World Academy of Sciences (TWAS), Italia

International Center for Genetic Engineering and Biotechnology (ICGEB), Italia

British Council (Reino Unido) – CONICOR, Córdoba, Argentina

European Communities (EC, Europe) 1995-1997

Howard Hughes Medical Institute (HHMI), EEUU

Association Francaise contre les Myopathies (AFM), Francia

The FSH Society, EEUU

PUBLICACIONES CIENTIFICAS

de Mendoza, D. and **Rosa, A.L. (1985)**. Cloning of mini-Mu bacteriophage in cosmids. *In vivo* packaging into phage lambda heads. **Gene** 39, 55-59.

de Mendoza, D, Gramajo, H. and **Rosa, A.L. (1986)**. In vivo cloning of DNA into multicopy cosmids by mini-Mu-cosduction. **Mol. Gen. Genet.** 205, 546-549.

Rosa, A.L. and Maccioni, H.J.F. (1987). Clathrin coated vesicles in *Neurospora crassa*. **Mol. Cell. Biochem.** 77, 63-70.

Rosa, A.L., Alvarez, M.E., Lawson, D. and Maccioni, H.J.F. (1990). A polypeptide of 59 kDa is associated with bundles of cytoplasmic filaments in *Neurospora crassa*. **Biochem. J.** 268, 649-655.

Rosa, A.L., Peralta-Soler, A. and Maccioni H.J.F. (1990). Purification of P59Nc and immunocytochemical studies of the 8-10 nm cytoplasmic filaments from *N. crassa*. **Exp. Mycol.** 14, 360-371.

Rosa, A.L., Alvarez, M.E. and Maccioni, H.J.F. (1990). Abnormal cytoplasmic bundles of 8-10 nm filaments in the *Neurospora crassa* snowflake colonial mutant contain P59Nc. **Exp. Mycol.** 14, 372-380.

Alvarez, M.E., **Rosa, A.L.**, Daniotti, J.L., Maggi, R. and Maccioni, H.J.F. (1991). Antibodies against the polypeptide of the *N. crassa* 8-10 nm filaments immunodetect a 59 kDa polypeptide in specialized rat epithelial cells. **Mol. Cell. Biochem.** 106, 125-131.

Haedo, S., Temporini, E., Alvarez, M.E., Maccioni, H.J.F. and **Rosa, A.L.** (1992). Molecular cloning of a gene (*cfp*) encoding the cytoplasmic filament protein P59Nc and its genetic relationship to the snowflake locus in *Neurospora crassa*. **Genetics** 131, 575-580.

Haedo, S., Mautino, M. and **Rosa, A.L.** (1992). Non Oak Ridge RFLP patterns in the *Neurospora crassa* multicent-2 strain. **Fungal Genet. Newslett.** 39, 23.

Temporini, E. and **Rosa, A.L.** (1993). Pleiotropic and differential phenotypic expression of two *sn* (snowflake) mutant alleles in *Neurospora crassa*: analyses in homokaryotic and heterokaryotic cells. **Curr. Genet.** 23, 129-133.

Alvarez, M.E., **Rosa, A.L.***, Temporini, E., Panzetta, G., Patrito, L., Wolstenholme, A. and Maccioni, H.J.F. (1993). The 59-kDa polypeptide constituent of 8-10-nm cytoplasmic filaments in *Neurospora crassa* is a pyruvate decarboxylase. **Gene** 30, 253-258. *(corresponding author)

Mautino, M., Haedo, S. and **Rosa, A.L.** (1993). Physical mapping of meiotic crossover events in a 200 kb cloned region of *Neurospora crassa* Linkage Group I. **Genetics** 134, 1077-1083.

Tazebay, U., Sophianopoulou, V., **Rosa, A.L.**, Scazzocchio, C. and Diallinas, G. (1994). Structure-function analysis of the proline permease (PRNB) of the filamentous fungus *Aspergillus nidulans*. **Folia Microbiol.** 39, 551.

Mautino, M.R., Goyon, C. and **Rosa, A.L.** (1996). Cloning and sequence of the *Ascobolus immersus* S-adenosyl-methionine synthetase-encoding gene. **Gene** 170, 155-156

Mautino, M.R., Barra, J.L and **Rosa, A.L.** (1996). *eth-1*, the *Neurospora crassa* locus encoding S-adenosylmethionine synthetase: molecular cloning, sequence analysis and *in vivo* overexpression. **Genetics** 142, 789-800

Barra, J.L., Mautino, M.R and **Rosa, A.L.** (1996). A dominant negative effect of *eth-1'*, a mutant allele of the *Neurospora crassa* S-adenosylmethionine synthetase encoding-gene conferring resistance to the methionine toxic analogue ethionine. **Genetics** 144, 1455-1462

Rosa, A.L., Haedo, S.D., Temporini, E.D., Borioli, G.A. and Mautino, M.R. (1997). Mapping chromosome landmarks in the centromere I region of *Neurospora crassa*. **Fungal Genet. Biol.** 21, 315-322

Morfini, G., Quiroga, S., **Rosa, A.L.**, Kosik, K. and Cáceres, A. (1997). Suppression of KIF2 in PC12 cells alters the distribution of a growth cone nonsynaptic membrane receptor and inhibits neurite extension. **J. Cell Biol.** 138, 657-669

Mautino, M.R. and **Rosa, A.L. (1998)**. Analysis of models involving enzymatic activities for the occurrence of C-T transition mutations during repeat-induced point mutation (RIP) in *Neurospora crassa*. **J. Theor. Biol.** 7, 61-71

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Temporini, E.D., Alvarez, M.E., Mautino, M.R., Folco, H.D., **Rosa, A.L. (2004)** The *Neurospora crassa cfp* promoter drives a carbon-source-dependent expression of transgenes. **J. Appl. Microbiology** 96, 1256-1264.

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Larovere, L., Romero, N., Fairbanks, L.D., Conde, C., Guelbert, N., **Rosa, A.L.**, Dodelson de Kremer R. (2004) A novel missense mutation, c.584A>C (Y195S), in two unrelated Argentine patients with hypoxanthine–guanine phosphoribosyl- transferase deficiency, neurological variant. **Mol. Genet. Metab.** 81, 352-354.

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Tenorio J, Alarcón P, Arias P, Dapía I, García-Miñaur S et al. (SOGRI Consortium: **Rosa AL** et al.) (2020) Further delineation of neuropsychiatric findings in Tatton-Brown-Rahman syndrome due to disease-causing variants in DNMT3A: seven new patients. **Eur J Hum Genet.** 28:469-479. doi: 10.1038/s41431-019-0485-3

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PREMIOS

Beca Marie Curie Commission European Communities. Bruxelas, Holanda. Institut de Genétique et Microbiologie, Université Paris-Sud (Francia) (1994)

Premio “**Diez Jóvenes Sobresalientes del Año**”, Premio anual de la Bolsa de Comercio de Córdoba, Argentina (1994)

Premio Universidad, Primer premio, categoría Profesores. Universidad Nacional de Córdoba, Argentina (**1992 y 1994**)

Beca Senior Scientist Poste Vert Institut National de la Recherche Scientifique (INSERM) - Hôpital Necker - Enfants Malades (Paris, Francia) (**1996-1997**)

Howard Hughes Medical Institute (USA), Fellow/Grant – International Program Laboratorio Genética y Biología Molecular INIMEC – CONICET, Córdoba (Argentina) (**1998-2003**)

Córdoba, Octubre 2022